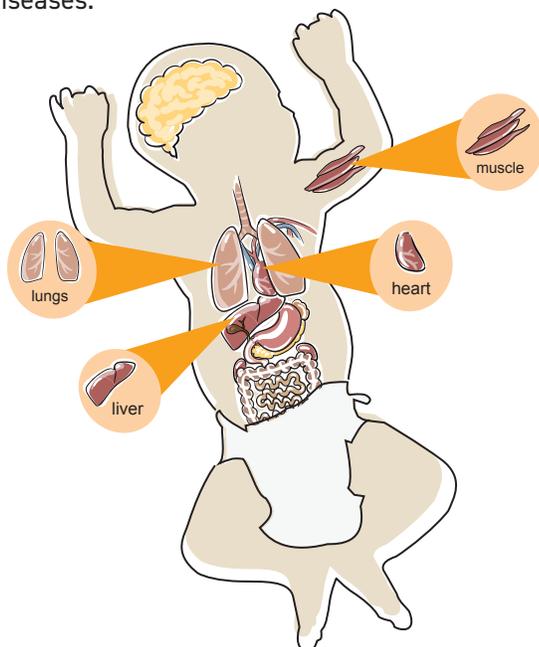


IOPD is a rapidly progressive disorder that can present in the first few days to months of life, often characterized by severe muscle weakness, cardiac disease, and respiratory involvement. IOPD can present less rapidly sometimes with less severe cardiac involvement. These patients can also present with early signs of muscle weakness and delayed motor milestones. IOPD patients who present with non-classic symptoms are a bit more difficult to diagnose because the signs and symptoms are variable and can overlap with other diseases.



The following table describes medical terms you may hear from your doctor or come across while reading about Pompe disease. For certain potentially unfamiliar medical terms, further description is found in the right-hand column. If you have a child or a family member who is experiencing any of these symptoms, consider talking to your doctor about Pompe disease. Please note: this is a brief summary and not an all-inclusive list of the signs and symptoms of IOPD.

System and symptoms	Definition
Cardiovascular (heart)	
Cardiomegaly	Enlarged heart
Hypertrophic cardiomyopathy	Larger than normal heart muscle due to thickening (bigger is not better when it comes to the heart muscle)
Arrhythmias	Changes in speed and rhythm of the heartbeat, can be fast or slow
Breathing (respiratory)	
Respiratory muscle weakness (including diaphragm, intercostal, and accessory muscles)	Weakness in the muscles used for breathing including the diaphragm and the muscles between the ribs
Respiratory distress	Difficulty breathing that may be due to frequent upper respiratory tract infections or pneumonia; can lead to respiratory failure

System and symptoms	Definition
Respiratory failure	Abnormal levels of oxygen or carbon dioxide in the blood due to difficulties breathing
Sleep apnea	Interrupted breathing during sleep
Exertional dyspnea	Shortness of breath after exercise
Orthopnea (indicative of early involvement of diaphragm)	Shortness of breath while lying down (supine) versus an upright position due to diaphragmatic weakness (the muscles used for breathing)
Weak cry	Cry that seems less robust than expected
Neurologic/musculoskeletal	
Hypotonia	Low muscle tone; may be called “floppy baby syndrome”
Generalized muscle weakness (proximal muscles most severely affected)	Weakness in the proximal muscles (shoulders, upper arms, pelvic area, and thighs) and distal muscles (lower arms and legs, hands, and feet)
Limb-girdle weakness	Weakness in the upper arms and legs
Neck (due to poor head control)	
Trunk muscles	<ul style="list-style-type: none"> • Proximal muscles: upper and lower extremities affected equally • Distal muscles: lower extremities slightly more affected than upper
Facial muscle myopathy with macroglossia	Weakness in the facial muscles causing an open mouth posture and an enlarged tongue that often protrudes
Absence, delay, or regression of motor milestones	Failure to reach, or reversal of, the usual age-appropriate childhood milestones in muscle development, strength, and tone
Calf muscle hypertrophy	Enlarged calf muscles
Delayed milestones	Failure to reach the usual age-appropriate childhood milestones
Exercise intolerance	Decreased ability to perform exercises at what would be considered a normal level or duration
Myalgia	Muscle pain
Low back pain	Pain in lower back
Scoliosis	Abnormal curvature of the spine
Atrophy (especially proximal muscles)	Decrease in muscle mass (also called muscle wasting)
Rigid spine syndrome	Progressive contracture and weakness in the muscles along the spine resulting in abnormal posture and limitations in neck/back movement
Ambulation difficulties	Difficulty walking

System and symptoms	Definition
Ptosis	Drooping of the eyelids
Fatigue	Lack of energy or feeling of exhaustion; can be physical, mental, or both, and may not be relieved by rest
Poor reflexes (in the later stages of the disease)	Weak or absent response to stimulus
Gastrointestinal (stomach, intestines, digestion/nutrition)	
Hepatomegaly	Enlarged liver
Failure to thrive	Weight and height measurements that fall below what is expected for a particular age and gender
Macroglossia	Enlargement of the tongue
Poor suck reflex, feeding and swallowing difficulties	Failure of an infant to suck in response to oral stimulation, difficulty with feeding and swallowing
Other	
Hearing impairment	Difficulty hearing or hearing loss